



congenital deafness with labyrinthine aplasia, microtia, and microdontia

Congenital deafness with labyrinthine aplasia, microtia, and microdontia (also called LAMM syndrome) is a condition that affects development of the ears and teeth. In people with this condition, the structures that form the inner ear are usually completely absent (labyrinthine aplasia). Rarely, affected individuals have some underdeveloped inner ear structures in one or both ears. The abnormalities of the inner ear cause a form of hearing loss called sensorineural deafness that is present from birth (congenital). Because the inner ear is important for balance as well as hearing, development of motor skills, such as sitting and crawling, may be delayed in affected infants. In addition, people with LAMM syndrome often have abnormally small outer ears (microtia) with narrow ear canals. They can also have unusually small, widely spaced teeth (microdontia).

Frequency

LAMM syndrome is a rare condition, although its prevalence is unknown. Approximately a dozen affected families have been identified.

Genetic Changes

LAMM syndrome is caused by mutations in the *FGF3* gene, which provides instructions for making a protein called fibroblast growth factor 3 (FGF3). By attaching to another protein known as a receptor, the FGF3 protein triggers a cascade of chemical reactions inside the cell that signal the cell to undergo certain changes, such as dividing or maturing to take on specialized functions. During development before birth, the signals triggered by the FGF3 protein stimulate cells to form the structures that make up the inner ears. The FGF3 protein is also involved in the development of many other organs and structures, including the outer ears and teeth.

FGF3 gene mutations involved in LAMM syndrome alter the FGF3 protein. The altered protein likely has reduced or absent function and is unable to stimulate signaling. The loss of FGF3 function impairs development of the ears and teeth, which leads to the characteristic features of LAMM syndrome.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- congenital deafness with inner ear agenesis, microtia, and microdontia
- deafness with LAMM
- LAMM syndrome

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Deafness with labyrinthine aplasia microtia and microdontia (LAMM)
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1853144/>

Other Diagnosis and Management Resources

- GeneReview: Congenital Deafness with Labyrinthine Aplasia, Microtia, and Microdontia
<https://www.ncbi.nlm.nih.gov/books/NBK100664>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Living with Hearing Loss
<https://medlineplus.gov/ency/patientinstructions/000360.htm>
- Health Topic: Ear Disorders
<https://medlineplus.gov/eardisorders.html>

- Health Topic: Hearing Disorders and Deafness
<https://medlineplus.gov/hearingdisordersanddeafness.html>
- Health Topic: Hearing Problems in Children
<https://medlineplus.gov/hearingproblemsinchildren.html>

Genetic and Rare Diseases Information Center

- Deafness with labyrinthine aplasia microtia and microdontia (LAMM)
<https://rarediseases.info.nih.gov/diseases/10707/deafness-with-labyrinthine-aplasia-microtia-and-microdontia-lamm>

Additional NIH Resources

- National Institute on Deafness and Other Communication Disorders
<https://www.nidcd.nih.gov/>

Educational Resources

- Centers for Disease Control and Prevention: Hearing Loss in Children
<https://www.cdc.gov/ncbddd/hearingloss/>
- KidsHealth from Nemours: Your Ears
<http://kidshealth.org/en/kids/ears.html>
- Orphanet: Deafness with labyrinthine aplasia, microtia, and microdontia
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=90024

Patient Support and Advocacy Resources

- Resource List from the University of Kansas Medical Center: Hard of Hearing/Deafness
<http://www.kumc.edu/gec/support/hearing.html>

GeneReviews

- Congenital Deafness with Labyrinthine Aplasia, Microtia, and Microdontia
<https://www.ncbi.nlm.nih.gov/books/NBK100664>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?term=%22congenital+deafness+with+labyrinthine+aplasia%2C+microtia%2C+and+microdontia%22+%5BDISEASE%5D+OR+NCT00023049+%5BID-NUMBER%5D+OR+NCT00400413+%5BID-NUMBER%5D+OR+NCT00802425+%5BID-NUMBER%5D>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28congenital+deafness+with+labyrinthine+aplasia,+microtia,+and+microdontia%29+OR+%28LAMM+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

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Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18435799>

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